MCEE gene

methylmalonyl-CoA epimerase

Normal Function

The *MCEE* gene provides instructions for making an enzyme called methylmalonyl CoA epimerase, which converts one form of the molecule methylmalonyl CoA to another. Specifically, the enzyme converts D-methylmalonyl CoA to L-methylmalonyl CoA. This conversion takes place within the pathway that converts the molecule propionyl-CoA to succinyl-CoA. This pathway is important in the breakdown of certain protein building blocks (amino acids), specific fats (lipids), and cholesterol.

Health Conditions Related to Genetic Changes

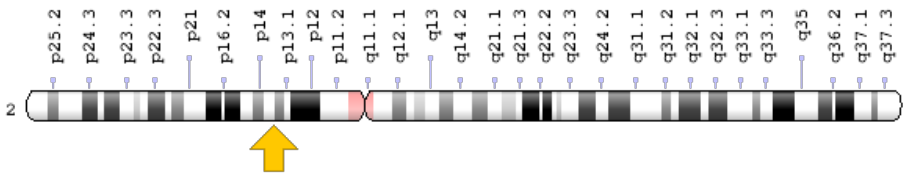
methylmalonic acidemia

At least three mutations in the *MCEE* gene have been found to cause methylmalonic acidemia, a condition characterized by feeding difficulties, developmental delay, and long-term health problems. These mutations are thought to result in the production of a methylmalonyl CoA epimerase enzyme with little or no function. People with methylmalonic acidemia caused by mutations in the *MCEE* gene typically have milder signs and symptoms than people with the condition caused by mutations in other genes. The features may be milder because there is an alternate pathway for the conversion of propionyl-CoA to succinyl-CoA that does not involve methylmalonyl CoA epimerase, so some succinyl-Co is produced even when there are mutations in the *MCEE* gene. This alternate pathway cannot compensate for the breakdown of certain molecules that occurs in the regular pathway, so people with *MCEE* gene mutations still have a buildup of the byproducts of some amino acids and certain fats. As a result, these toxic compounds build up in the body's organs and tissues, causing the signs and symptoms of methylmalonic acidemia.

Chromosomal Location

Cytogenetic Location: 2p13.3, which is the short (p) arm of chromosome 2 at position 13.3

Molecular Location: base pairs 71,109,676 to 71,130,288 on chromosome 2 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- DL-methylmalonyl-CoA racemase
- GLOD2
- glyoxalase domain containing 2
- MCEE_HUMAN
- methylmalonyl CoA epimerase
- methylmalonyl-CoA epimerase, mitochondrial
- methylmalonyl-CoA epimerase, mitochondrial precursor

Additional Information & Resources

GeneReviews

- Isolated Methylmalonic Acidemia
  https://www.ncbi.nlm.nih.gov/books/NBK1231

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28MCEE%5BTIAB%5D%29+OR+%28methylmalonyl+CoA+epimerase%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- METHYLMALONYL-CoA EPIMERASE
  http://omim.org/entry/608419

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=MCEE%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=16732
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/84693
- UniProt
  http://www.uniprot.org/uniprot/Q96PE7

Sources for This Summary

- Bikker H, Bakker HD, Abeling NG, Poll-The BT, Kleijer WJ, Rosenblatt DS, Waterham HR, Wanders RJ, Duran M. A homozygous nonsense mutation in the methylmalonyl-CoA epimerase gene (MCEE) results in mild methylmalonic aciduria. Hum Mutat. 2006 Jul;27(7):640-3.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16752391
- Dobson CM, Gradinger A, Longo N, Wu X, Leclerc D, Lerner-Ellis J, Lemieux M, Belair C, Watkins D, Rosenblatt DS, Gravel RA. Homozygous nonsense mutation in the MCEE gene and siRNA suppression of methylmalonyl-CoA epimerase expression: a novel cause of mild methylmalonic aciduria. Mol Genet Metab. 2006 Aug;88(4):327-33. Epub 2006 May 11.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16697227
- Gradinger AB, Bélair C, Worgan LC, Li CD, Lavallée J, Roquis D, Watkins D, Rosenblatt DS. Atypical methylmalonic aciduria: frequency of mutations in the methylmalonyl CoA epimerase gene (MCEE). Hum Mutat. 2007 Oct;28(10):1045.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17823972
- OMIM: METHYLMALONYL-CoA EPIMERASE
  http://omim.org/entry/608419

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https://ghr.nlm.nih.gov/gene/MCEE

Reviewed: July 2011
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
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